Gene News

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What's Happening?

In short...a lot. With three active grants in addition to our regular duties, we are anticipating a busy and productive time for public health genetics. We are in the second and final year of funding on the Hawaii Genetic Assessment and Planning Project and are in the first year of funded activities on two new grants (see page 2 for details), we look forward to collaborating with other DOH programs and the community in keeping the "Health State" tradition.

New Faces

The Genetics Program welcomes Nicole Sameit into the Children with Special Health Needs (CSHN) Ohana as an Assistant Project Coordinator for the Gene AID Project. We also want to welcome Sharon Hirose who recently joined our program as a clerk. Nicole and Sharon join Sylvia Au, the State Genetics Coordinator.

New Beginnings

It's been a several years since the Genetics Program had a regular newsletter. However, with the addition of new staff, we are happy to bring it back and keep you in touch with what is happening.

HGAPP UPDATE

In the second year of the Hawaii Genetic Assessment and Planning Project (HGAPP), we are close to successfully completing our goals of expanding the current state genetic needs assessments and developing a revised State Genetics Plan. Since the last needs assessment in 1993, there have been significant changes in medicine, technology, and policy. HGAPP expands the needs assessment activities to address the future concerns associated with the new sciences and technologies spurred by the advancements in research and medicine.

During the past year, we have interviewed and surveyed physicians, public health nurses and administrators. We are also completing our focus groups with members of the general public. The focus group responses have been quite interesting and helpful.

In 2002, we will use the needs assessment data and other national guidelines and recommendations to draft our revised State Genetics Plan. We hope to have this outline for your review, comments, and recommendations by April 2002. A public forum will be held to discuss the proposed Plan in June 2002.

To view the current State Genetics Plan, go to www.hawaiigenetics.org or contact our office for a copy if you do not have internet access.

Inside this issue:

Did you know that humans and bananas share 50% of our genomes?

Hawaii Department of Health
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Hawaii Genetic Awareness, Implementation, and Data Project

The Hawaii Genetic Awareness, Implementation, and Data Project (Gene AID), is a three year federal HRSA grant funded project. Gene AID is designed to help integrate the Newborn Metabolic Screening Program (NBMSP), Newborn Hearing Screening Program (NBHSP), Birth Defects Program (BDP), and Early Intervention Services data systems within the framework of federal and state laws. The Project will also develop activities to increase genetic awareness for health care providers, public health programs, policy makers, school age children, and other broad audiences.

Activities associated with increasing genetic awareness and genetic education include: creating a lecture series (the preferred method of education from our preliminary needs assessment results), educational workshops, and conferences for health care providers; providing web based education modules for teachers; and developing fun learning activities for the broader community. The completion of the Human Genome Project and the changing face of medicine, make it essential that people in Hawaii have a basic awareness of the new advances in genetics.

Contact Information:

Sylvia Au: 733-9063 sylvia@hawaiigenetics.org

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Tandem Mass Spectrometry: Financial, Ethical, Legal, and Social Issues Project

Tandem Mass Spectrometry is a technology that allows expansion of our current newborn screening panel of 7 disorders to 30+ additional disorders without a great additional cost. However promising, new technologies often create problems that must be identified, addressed, and resolved in order for operations to run smoothly in an ethical, fair, and culturally competent program.

The MS/MS project is another HRSA federally funded three year multi-state grant project. It is designed to research, identify strategies, and develop materials for addressing the ethical, legal, social, and financial issues surrounding the use of Tandem Mass Spectrometry for newborn screening of culturally and ethnically diverse populations. The Hawaii Genetics Program is the lead for the project that also includes California, Oregon, Washington, Alaska, and Idaho.

June 26th, 2001 marked the one year anniversary of the Human Genome Project and Celera Genomics' joint announcement that a "working draft" of the Human Genome had been completed.

Tandem Mass Spectrometry: A brief explanation

A Tandem Mass Spectrometer (MS/MS) is one of several types of analytical instruments known as a mass spectrometer that measures the weights of molecules. A Tandem Mass Spectrometer can be thought of as two machines in a series connected by a chamber that measure the level of compounds in newborns blood called amino acids and acylcarnitines. Too much of these compounds in the blood may indicate that the child has inherited a metabolic disorder and requires diagnostic testing.

The MS/MS technology is very precise and should greatly reduces the number of false positives and false negatives for diseases like PKU. One of the benefits of MS/MS technology is the ability to measure more than one compound simultaneously in a single, two minute analysis. Currently this technology can detect over 30 disorders of inborn metabolism.

Birth Defects in Hawaii

True or False? In Hawaii …

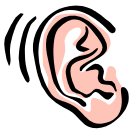
1) 1 in 5 pregnancies (20%) result in a miscarriage.
2) About 1,000 (5%) of all babies are born each year with some recognizable abnormality.
3) About 30% of all pediatric hospital admissions are a result of problems from birth defects.
4) Birth defects are the #1 cause of infant mortality.
5) Some birth defects are completely preventable.

If you answered **"True"** to each of the above, you are absolutely correct. A birth defect is any structural, functional or biochemical abnormality in development that originates before birth and is detectable at birth or shortly thereafter. These abnormalities may be genetic, caused by environmental hazards or adverse life style effects, but often are of unknown origin.

In response to this core public health concern, the Hawaii Birth Defects Program (HBDP) was established in 1988 as a statewide data surveillance system to provide valid, reliable and timely information on the number of pregnancies and children affected by these defects, and for assessing the effectiveness of control programs aimed at reducing the number of birth defects and other adverse reproductive outcomes.

The HBDP monitors birth defects for trends and changes over time, including the identification of geographic and other clusters. The Program distributes data/information, gives presentations, and publishes articles. It carries out special studies, and provides data for developing public awareness programs about birth defects and their causes. And lastly, the HBDP acts as an information source for the planning and development of appropriate statewide and community level services and preventive strategies.

Data and information from years 1986-1999 for over 15,000 birth defect cases can be obtained from the Hawaii Birth Defects Program, 620 Waipa Lane, #206, Honolulu, Hawaii 96817-3533, Tel: 808-832-0278, Fax:808-832-0288, E-Mail: hbdp@crch.hawaii.edu.



Now Hear This…

On July 1, 2001, the Act to establish mandatory hearing impairment screening for newborns and infants in the state of Hawaii was revised. The revised statute mandates newborn hearing screening, much like our State's mandated newborn metabolic screening. This mandatory screening enables the early identification of children with hearing loss and the promotion of their development of language and communication skills.

The next challenge for the Newborn Hearing Screening (NHSP) staff will be to establish standards and guidelines for screening, identification, diagnosis, intervention, and monitoring of children at risk for or with confirmed hearing loss. If you have any questions about the NBHS Program, please call the Program Coordinator, Yusnita Weirather, at 973-1115.

Humans have 99.9% of their genetic information identical to each other.

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Newborn Metabolic Screening Update

Since July 1, 1997, all newborn specimens from Hawaii are being sent to the Oregon State Public Health Laboratory for the testing of seven disorders: phenylketonuria (PKU), congenital hypothyroidism, galactosemia, hemoglobinopathies, congenital adrenal hyperplasia (CAH), biotinidase deficiency and maple syrup urine disease (MSUD). State law requires that all newborns be screened for these seven disorders, which can cause severe mental and growth retardation or even death if not treated early in the newborn period.

In July 2001, Hawaii began collaborating with the state of California on a 3 year pilot program to study the laboratory and clinical reliability, validity, and utility of Tandem Mass Spectrometry which can detect 30+ inborn errors of metabolism. Contact Chris Matsumoto at 733-9069 or chris@hawaiigenetics.org for more information.

In the 4 1/2 years since Newborn Screening has been expanded in Hawaii, 76,689 newborns have been screened and more than 40 have been diagnosed with one of the seven disorders.

Disorder Screened For	Number of infants identified since 1997
Phenylketonuria	1
Congenital Hypothyroidism	28 (primary cases)*
Galactosemia	6 Duarte variants (0 classical)
Hemoglobinopathies	3 Sickle Cell Disease
Congenital Adrenal Hyperplasia	3
Biotinidase Deficiency	2*
Maple Syrup Urine Disease	3*

*** Hawaii has a higher incidence than the national average for congenital hypothyroidism (National: 1: 4,000; Hawaii: 1:2,739), MSUD (National: 1:150,000; Hawaii: 1:25,563), and biotinidase deficiency (National: 1:60,000; Hawaii 1:38,344).**